Reviewer's report

Title: A de novo complete BRCA1 gene deletion identified in a Spanish woman with early bilateral breast cancer.

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Reviewer: Thomas Hansen

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In this study, the author’s describe a de novo BRCA1 whole gene deletion in a woman with early bilateral breast cancer. Her family is without a previous history of breast or ovarian cancer. They analysed the BRCA1/BRCA2 genes by direct sequencing and MLPA analysis. The BRCA1 whole gene deletion was verified by confirmatory MLPA as well as array CGH. Their findings add to the current knowledge of BRCA1/BRCA2 de novo mutations and support the use of MLPA in individual’s sequenced negative for BRCA1/BRCA2 mutations.

The laboratory work is well performed and the results are well presented.

Minor Essential Revisions

1) The manuscript needs careful editing throughout, e.g. abstract, line 5, “analized” should be changed to “analysed”; CGH array should be changed to array CGH; page 5, line 9 “presented a no previous” should be changed to “presented no previous”; page 11, line 12 “which seem result” should be changed to “which seem to result” amongst others.

2) The nomenclature should be changed throughout the manuscript, e.g. c.744+14C>T should be changed to nucleotide 744+14C>T, since “c.” refers to the coding DNA sequence, where nucleotide +1 is the A of the ATG translation initiation codon. Moreover it should be mentioned that this mutation is seen in the homozygote state. Finally this mutation is not undescribed (described in UMD-BRCA2 mutation database), but uncharacterized.

3) Page 13, line 24, reference 43 should be included.

4) I do not understand why the authors use the term “mosaicism” on page 10, line 11 and page 11, line 16.

5) Does the symptoms caused by defects in the ZFPM2 gene mentioned on page 11, line 19-21 correlates to a duplication of this gene?

Level of interest: An article of importance in its field

Quality of written English: Needs some language corrections before being published
Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

None